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59-year-old female with breathlessness.

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A 59 year-old female underwent an ECG and echocardiographic screening. Her brother died at a quite young age of kidney failure. Resting ECG showed borderline voltage criteria for left ventricular hypertrophy, with marked widespread T wave inversion. Echocardiogram was normal, but in consideration of exertional breathlessness and abnormal baseline ECG, she underwent a coronary angiogram which showed unobstructed coronaries. She was then referred to have a cardiac magnetic resonance (CMR) for further characterization. CMR images were acquired with a 1.5 Tesla scanner and the imaging protocol included SSFP cine images (Figure 1A) as well as late gadolinium enhancement images (LGE) in the long- and short-axis planes covering the whole left ventricle (Figure 1B). In addition, native and post-contrast T1-mapping (MOLLI) images were acquired for further tissue-characterization (Figure 1C and Figure 1D, respectively).

What is the most likely diagnose, based on CMR findings?

- A. Anderson-Fabry's disease (AFD)
- B. Cardiac amyloidosis
- C. Genotype(+), phenotype(-) hypertrophic cardiomyopathy (HCM)
- D. Myocardial iron overload
- E. Normal heart

Answer A

The wall thickness was normal (Figure 2A). However, this per se does not exclude all the above possible diagnosis, as early phase disease might be present. Post-contrast images showed mid-wall myocardial LGE in the basal inferolateral wall (Figure 2B, white arrow). The native T1 mapping images demonstrate diffusely reduced T1 values, with the exception of the basal inferolateral wall where the values were pseudo-normal (Figure 2C, white arrow).

Given the presence of LGE option E should be excluded, as LGE is not a normal finding.

Option B and C can also be excluded because in both cardiac amyloidosis and HCM the native T1 mapping is normally increased¹⁻³.

Option D can be compatible with low native T1 values but it not typical to find in these patients the LGE and LGE with this pattern.

Post-contrast T1-mapping values were diffusely reduced in all myocardial segments and even lower in the basal and mid-cavity inferolateral wall (Figure 2D arrow), correlating with the presence of LGE. Low native T1-mapping values, together with pseudonormalization of these in the inferolateral wall, have been recently described as pathognomonic of AFD^{4,5}.

The diagnosis was confirmed by the detection of low dosage of leucocyte α -galactosidase activity. AFD is an X-linked storage disease caused by deficiency in the enzyme α -galactosidase A. Female patients have been shown to have incomplete phenotypes, most commonly the presence of LGE without LVH⁵. Treatment with recombinant enzyme is available. Our case shows how new CMR relaxometry techniques offer an opportunity for early diagnosis.

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